Preparation for Term Paper (Height QTL Articles)

Human Height Publication
([Yengo et al. 2022](#))

- Abstract indicates that 12,111 independent SNPs covering 21% of the genome account for nearly all of common SNP-based heritability.

- Introduction references [Yang et al. 2010](#) as an earlier height GWAS.
  - Along with this publication, tracking citations may help with identifying other relevant publications.
  - In the discussion, it is mentioned that the sample size for this study is "*more than seven times that of previous studies*."

- Introduction also includes a warning that a sufficiently large sample size could implicate the entire human genome (referencing [Flint and Ideker 2019](#)), and defines "saturation" for a nearly complete or "saturated" collection of common variation (defined in various ways, but with consistent conclusions).
  - For example, SNP-based heritability of height is defined by Genome-Wide Significant (GWS) SNPs explain ~100% of such variability in European ancestry individuals and >90% of such variability in non-European ancestry individuals.
  - All HapMap3 (HM3) SNPs are also considered, combining with GWS SNPs to define total SNP heritability.
  - While perhaps outside of the scope for this class / project, I also noticed [Barry et al. 2023](#) describing different ways to estimate heritability. In general, I think this may be a topic of some importance, and I am interested to see the effect of the methodology on the QTL analysis.

- Prediction accuracy (R^2) for PolyGenic Scores (PGS) is always less than 50%
  - For comparison, the **Parental Average (PA)** for height has a prediction accuracy of 43.8% in *Figure 4b*, with an overlapping interval for PGS from GWS SNPs or HM3 SNPs.
  - With either GWS or HM3 SNPs, the combination of PA *and* PGS has an R^2 of approximately 55%

- My understanding is that meta-analysis is performed, where analysis methods may vary from when individual variant information is available for a QTL.

Maize SLAF-Seq QTL Publication
([Fei et al. 2021](#))

- Introduction / Background mentions using *MaizeGDB* as resource to find QTL related to plant height and ear height.

- *ICIMapping 4.2* was used to define a genetic map.

- *HighMap* is also used for analysis. The provided reference [31] is for *KEGG*, but I believe the citation should be Liu et al. 2014.

- *R/qtl* is described as being used for QTL analysis.

- In general, I believe SLAF-Seq shares some similarity to ddRADseq, and I found a Sun et al. 2013 publication.
  - The methods in this publication describe selecting Simple Sequence Repeat (SSR) markers using *MaizeGDB*.

Brassica napus QTL Publication ([Dong et al. 2021](#))

- "Traditional" QTL analysis was performed along with QTL-Seq, and consistent results were considered for further interpretation.
  - In this study, the QTL-Seq consisted of two pools: the 20 tallest plants in the "T" pool and the 20 shortest ("dwarf") plants in the "D" pool.
  - I believe the "traditional" analysis was includes 200 Whole Genome Sequencing samples, and other data types were also considered.
- *HighMap* and *[R/qtl](#)* are mentioned in the Methods.
  - Genetic distance was reported to be calculated using the *Kosambi* map distance.
  - Use of the *SEgregate Analysis (SEA)* R-package is also described.
- There is a section titled "*Construction of a relatively* **saturated** *SNP-based genetic linkage map using WGS*".
  - This study focuses on a Whole Genome Sequencing method (with the 200 samples).
  - However, it is reported that this is "higher quality" than maps created with other markers (including SSR, RAD, and SLAF).
- There is a data deposit in [CNP0001630](#), but I unfortunately can't read all of the details associated with the data deposit.

Other Articles Related to Height QTL (or GWAS)

- Visscher et al. 2010 provide a history/review of studying human height.

- Hirschhorn and Lettre 2009 provide a review of GWAS for human height (up to the time of publication).

- Abdellaoui et al. 2023 is an example of a GWAS review (including some discussion related to height).
  - Tam et al. 2019, Caballero et al. 2015, and Cano-Gamez and Trynka 2020 are also examples of a general reviews.

- In general, Privé et al. 2022 describe impacts of "misspecification" on results.